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Leukaemia Section
Short Communication

dic(9;16)(p13;q11) PAX5/?

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Abstract

Short communication on dic(9;16)(p13;q11) PAX5/?, with data on clinics, and the genes implicated.

Clinics and pathology

Disease

Acute lymphoblastic leukemia (ALL)

Epidemiology

Only one case to date, a 15-year old boy with a pre-B-ALL (Coyaud et al., 2010).

Cytogenetics

Cytogenetics morphological

The dic(9;16) was the sole abnormality within a subclone.

Genes involved and proteins

PAX5

Location
9p13.2

Protein
391 amino acids; from N-term to C-term, PAX5 contains: a paired domain (aa: 16-142); an octapeptide (aa: 179-186); a partial homeodomain (aa: 228-254); a transactivation domain (aa: 304-359); and an inhibitory domain (aa: 359-391).

Lineage-specific transcription factor; recognizes the concensus recognition sequence GNCCANTGAAGCGTGAC, where N is any nucleotide. Involved in B-cell differentiation. Entry of common lymphoid progenitors into the B cell lineage depends on E2A, EBF1, and PAX5; activates B-cell specific genes and repress genes involved in other lineage commitments. Activates the surface cell receptor CD19 and repress FLT3. Pax5 physically interacts with the RAG1/RAG2 complex, and removes the inhibitory signal of the lysine-9-methylated histone H3, and induces V-to-DJ rearrangements. Genes repressed by PAX5 expression in early B cells are restored in their function in mature B cells and plasma cells, and PAX5 repressed (Fuxa et al., 2004; Johnson et al., 2004; Zhang et al., 2006; Cobaleda et al., 2007; Medvedovic et al., 2011).

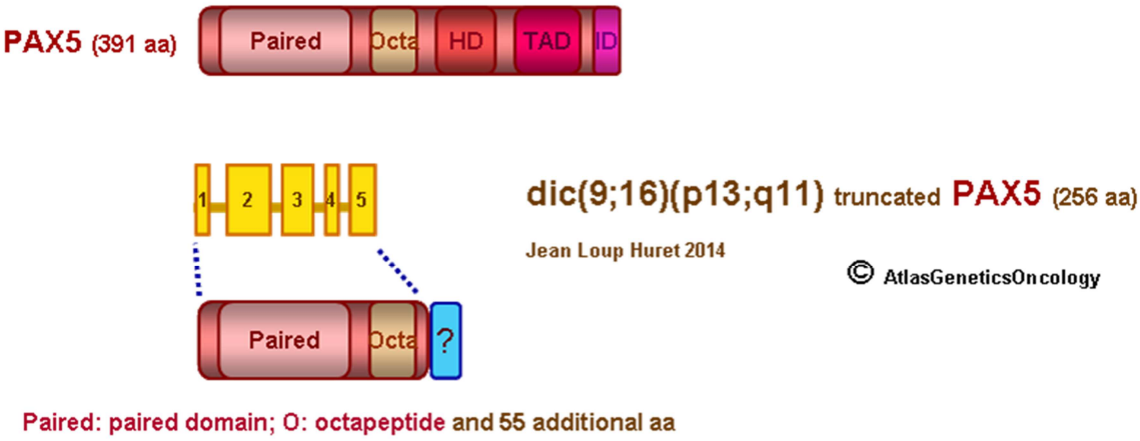
Result of the chromosomal anomaly

Hybrid gene

Description
Truncation of PAX5 after exon 5. The region in 16q11 does not contain any gene.

Fusion protein

Description
256 amino acids. The truncated protein contains the DNA binding paired domain and octapeptide of PAX5 (201 aa) and 55 aa from contiguous introns.



Truncated PAX5.

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